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Association of IL-4 -590 T/C and IL-4 receptor Q551R gene polymorphisms with vitiligo

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Background: Vitiligo is a depigmenting disorder with probable immune and genetic factors.

Objectives: This work aims to investigate the association between genetic polymorphisms related to interleukin 4 (IL-4) and IL4 receptor (IL-4R) genes with vitiligo.

Design and Setting: Case-control study conducted on 96 vitiligo Saudi patients recruited from dermatology clinics affiliated to Qassim University, Saudi Arabia. Thier data were compared with 87 normal healthy unrelated controls from the same locality. Genomic DNA was extracted and processed using the real time PCR amplification for characterization of polymorphisms related to IL-4 (-590 T/C) and IL-4R (Q551R A/G) genes.

Results: Vitiligo cases showed higher frequency of the mutant IL-4 (-590) CC genotype compared to controls, yet, statistically non-significant (61.2% vs. 53.8%, p>0.05). On the other hand, cases showed significantly higher frequency of the IL-4R (Q551R A/G) homozygous mutant polymorphism GG than in controls (22.3% vs. 8%, p=0.01, OR=3.3, 95% CI= 1.3-8.2) with a significantly lower frequency of the AG genotype (2.1% vs. 28.7%, p<0.0001). Allele frequency assessment showed a nonsignificant difference between cases and controls. Comparing case sub-groups related to demographic and clinical parameters showed also non significant difference regarding the frequency of all studied genetic variants (p>0.05).

Conclusion: This study provides an evidence for a significant association of IL-4R genetic polymorphisms with the susceptibility to vitiligo that needs further evaluation as being a useful genetic or pathophysiologic marker of the disease.